Day one-presentation-session I
Rachel Richesson

"Plans for data standards in rare disease registries"

Rachel Richesson

The inability to access existing standardized registry questions results in lengthy and resource-intensive registry development efforts and limits opportunities for data sharing. A standardized library of data elements will speed the development and deployment of patient registries and allow registries to share and receive data from other registries or data sources. In addition, a library of standardized registry questions will enable cross-indication and cross-disease analyses, facilitate collaboration, and generate more meaningful results for rare disease patients, physicians, and researchers. Rachel Richesson will briefly discuss a recently funded American Recovery and Reinvestment Act (ARRA) project that has implications for data standards and an infrastructure for registries across various rare disease. The primary objective of the Patient Registry Item Specifications and Metadata (PRISM) project is to develop standardized questions for patient registries across many different rare diseases, which can be used for developing new registries and revising existing ones. The library questions will use consistent structure and language, and the underlying data elements (i.e., questions + answers + definitions) will be encoded using data standards that will facilitate reliable and consistent data collection and enhance opportunities for question re-use and data sharing. Dr. Richesson will describe the PRISM project's standards strategy, project timeline, and future implications for registries across the spectrum of rare diseases.

*Describe ARRA project "Library of Standardized Patient Registry Questions for Rare Diseases"

-Standards Strategy
-Timeline
-Future implications for rare disease registries

Uniting Rare Diseases

Advancing Rare Disease Research:
The Intersection of Patient Registries, Biospecimen Repositories and Clinical Data

Session I
Standards, Informatics and Technology
January 11, 2009

Data Standards in Rare Disease Registries

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Where do I need to go

Welcome! You have reached the home page for the Rare Diseases Clinical Research Network (RDCRN). Each Consortium within the network provides detailed information on several rare diseases.

What if I am unsure of which consortium to visit?

Scan the list to the right for a disease name. Once you have located it, click on the link for a brief description which will lead you to the correct consortium.

How will this consortium be useful to me?

You can take action! Once you have reached the correct consortium, you will be able to **join the contact registry** for clinical research trials. You will also find several helpful resources that include participating clinical center information, support and advocacy group information and other useful links.

RDCRN Consortium Studies are opening! Learn More >> *Last Updated: 30 March*

Clinical Research Consortia :

Urea Cycle Disorders Consortium [Study Information]
- N-Acetylglutamate Synthase (NAGS) Deficiency
- Carbamyl Phosphate Synthetase (CPS) Deficiency
- Ornithine Transcarbamylase (OTC) Deficiency
- Argininosuccinate Synthetase Deficiency (Citrullinemia I)
- Citrin Deficiency (Citrullinemia II)
- Argininosuccinate Lyase Deficiency (Argininosuccinic Aciduria)
- Arginase Deficiency (Hyperargininemia)
- Ornithine Translocase Deficiency (HHH) Syndrome

Angelman, Rett, and Prader-Willi Syndromes Consortium [Study Information]
- Angelman Syndrome
- Rett Syndrome
- Prader-Willi Syndrome

CINCH - Consortium for Clinical Investigation of Neurological Channelopathies [Study Information]
- Andersen-Tawil Syndrome (a form of Periodic paralysis)
- Episodic Ataxias
- Non-dystrophic Myotonic Disorders

Bone Marrow Failure Disease Consortium [Study Information]
- Aplastic Anemia
- Myelodysplastic Syndromes
- Paroxysmal Nocturnal Hemoglobinuria (PNH)

Vasculitis Clinical Research Consortium [Study Information]
- Wegener's Granulomatosis (WG)
- Microscopic Polyangiitis (MPA)
- Churg-Strauss Syndrome (CSS)
- Polyarteritis Nodosa (PAN)
- Takayasu's Arteritis (TAK)
- Giant Cell (Temporal) Arteritis (GCA)

Rare Genetic Steroid Disorders Consortium [Study Informatio
- Congenital Adrenal Hyperplasia
- Androgen Receptor Defects
- Apparent Mineralocorticoid Excess (Low Renin Hypertension)

Rare Thrombotic Diseases Consortium [Study Information]
- Antiphospholipid Antibody Syndromes (APS)
- Heparin-induced Thrombocytopenia (HIT)
- Paroxysmal Nocturnal Hemoglobinuria (PNH)
- Catastrophic Antiphospholipid Antibody Syndrome (Thrombotic Stor
- Thrombotic Thrombocytopenic Purpura (TTP)

Rare Lung Diseases Consortium [Study Information]
- Hereditary Interstitial Lung Disease (hILD)
- Lymphangioleiomyomatosis (LAM)
- Pulmonary Alveolar Proteinosis (PAP)
- Alpha-1 Antitrypsin Deficiency (Alpha-1)

Genetic Diseases of Mucociliary Clearance Consortium [Stu

Patient Registry Item Specifications and Metadata …for Rare Diseases

Features

- Re-use
- Context of registries – vision, use-cases
- Identify appropriate standards / fill gaps
- Encourage best practices
- Indexing and retrieval of registry questions
- Model for growth and access of library
- Multi-disciplinary

Patient Registry Item Specifications and Metadata for Rare Disease Research

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